Claims

1. Purified nucleic acid comprising a human retinoblastoma gene, or a fragment thereof comprising 15 or more bases, said nucleic acid being less than 100kb in size.

2. A vector comprising the nucleic acid of claim 1.

3. A cell transformed with DNA encoding retinoblastoma polypeptide or a fragment thereof.

4. The nucleic acid of claim 1, wherein said nucleic acid hybridizes specifically to said retinoblastoma gene under hybridizing conditions.

5. An isolated polypeptide encoded by the nucleic acid of claim 1.

6. An antibody produced to the polypeptide of claim 5.

7. An antibody produced to naturally occurring retinoblastoma polypeptide.

8. A method of detecting large deletions in the retinoblastoma gene of a human patient predisposing said patient to retinoblastoma, comprising the steps of:

hybridizing a nucleic acid sample from said patient with a probe specific for the retinoblastoma gene, and

determining the ability of said probe to hybridize to said nucleic acid,

wherein lack of hybridization to said nucleic acid indicates the presence of a large deletion in said gene.

9. A method of detecting large deletions in the retinoblastoma gene of a human patient that may predisposing said patient to retinoblastoma, comprising the steps of:

generating nucleic acid fragments from a sample of said patient,

separating said fragments according to a determined physical property of said fragments,

hybridizing a probe specific for the retinoblastoma gene to said fragments,

detecting hybrids of said probe and said fragments, and

comparing said hybrids to hybrids detected from the hybridization of said probe and separated nucleic acid fragments from a normal retinoblastoma gene,

wherein the absence of hybrids, or the smaller size of said hybrids from the sample of said patient is an indication of large deletions in the retinoblastoma gene of said patient.

10. The method of claim 8 or 9, wherein the probe specific for the retinoblastoma gene is the cloned DNA in p4.7R, or a fragment thereof.

11. The method of claim 9, wherein the physical property is molecular weight.

12. A method of detecting small deletions or point mutations in the retinoblastoma gene of a human patient predisposing said patient to retinoblastoma, comprising the steps of:

determining the nucleotide sequence of a retinoblastoma allele, or subregion thereof, from said patient, and

comparing said nucleotide sequence with the nucleotide sequence of a retinoblastoma allele or region thereof from a person not afflicted with retinoblastoma.

13. A method of detecting small deletions or point mutations in the retinoblastoma gene of a human patient predisposing said patient to retinoblastoma, comprising detecting mismatches between a nucleic acid sample from said patient and a detectable probe specific for the retinoblastoma gene from a person not afflicted with retinoblastoma, wherein mismatches are an indication of small deletions or mutations in the retinoblastoma gene of said patient.

14. A method of diagnosing predisposition of a human patient to retinoblastoma, comprising detecting the co-inheritance of retinoblastoma alleles of said patient with DNA polymorphisms in a pedigree analysis.

15. A method of detecting genetic polymorphisms, in the retinoblastoma gene of a human patient, that predispose said patient to retinoblastoma, comprising the steps of:

generating nucleic acid fragments from a sample of said patient,

separating said fragments according to a determined physical property of said fragments,

hybridizing a detectable nucleic acid probe capable of hybridizing to the wild type retinoblastoma gene to said fragments,

detecting hybrids of said probe and said fragments, and

comparing said hybrids to hybrids detected from the hybridization of said probe and separated nucleic acid fragments from a sample of a parent of said patient,

wherein the co-inheritance of specific genetic polymorphisms with the retinoblastoma gene is an indication of the predisposition of said patient to retinoblastoma.

16. The method of claim 15, wherein said physical property is molecular weight.

17. A method of treating a human patient having a defective retinoblastoma gene comprising administering to said patient an anti-retinoblastoma-forming amount of the retinoblastoma polypeptide.

18. A composition suitable for treating a human patient having a defective retinoblastoma gene, comprising retinoblastoma polypeptide and a pharmacologically acceptable carrier therefor.

19. A method of detecting the presence, in a tumor sample, of a protein the absence of which is associated with a neoplasm, said method comprising producing an antibody to said protein, contacting said antibody with said tumor sample, and detecting immune complexes as an indication of the presence in said tumor sample of said protein.

20. A method of detecting the presence of the retinoblastoma protein in a tumor sample from a human patient, comprising the steps of:

contacting said tumor sample with an antibody which specifically reacts with the retinoblastoma protein, and

determining whether immune complexes are formed with said antibody, the formation of said immune complexes being an indication that the tumor is not retinoblastoma and the absence of immune complexes indicating that the tumor is retinoblastoma.

21. The method of claim 20, wherein said antibody is a monoclonal antibody.